There was no tendency for a correlation between this polymorphism and T1D in patients from Greece. In an attempt to clarify whether this polymorphism is also associated with other autoimmune diseases, further analyses based on Greek cohorts of patients are in progress. Functional studies will shed further light on this shared genetic pathway.

A71 THE CD40 REGION IS ASSOCIATED WITH MULTIPLE AUTOIMMUNE DISEASES IN THE GREEK POPULATION

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Background There is increasing evidence that different autoimmune diseases may share some common pathogenetic pathways. The CD40 locus has recently been identified by a genome-wide study as a genetic risk factor for rheumatoid arthritis (RA). CD40 is a member of the tumour necrosis factor receptor superfamily which is constitutively or inducibly expressed on the surface of a variety of immune and non-immune cell types, thus indicating that this locus might also be involved in other autoimmune diseases.

Aim To investigate whether the rs4810485 single nucleotide polymorphism (SNP) of CD40 is also associated with systemic lupus erythematosus (SLE), RA and type 1 diabetes (T1D) in the population of Greece.

Materials and Methods Genotyping of the variant defining the signal peak, rs4810485, was performed in Greek samples by PCR followed by electrophoretic analysis in a 2.2% agarose gel or using the Taqman assay (Applied Biosystems). The SLE, RA and T1D sample sets consisted of 382, 288 and 153 controls and 351, 272 and 48 patients, respectively. Patients and controls were sex- and age-matched. The statistical difference in genotype distribution and investigated variables was assessed by two-tailed χ^2 test and by Student unpaired t test. ORs and 95% CI were calculated.

Results The authors found that the risk allele G of the CD40 rs4810485 SNP, which has previously been implicated in the predisposition to RA in other populations, was more common in individuals with SLE and RA than in controls (p<0.0001, OR=77, 95% CI 1.42 to 2.2 and p=0.03, OR=31, 95% CI 1.02 to 1.7 respectively), thus concluding that the polymorphism examined is associated with the development of these diseases in our population. In contrast, no statistically significant difference in G or T allele (p>0.05) frequencies was observed between T1D patients and controls (p=0.00, OR=0.00, 95% CI 0.63 to 1.6). Replication of the study was performed using another T1D cohort originating from another geographical region of Greece but again no association was detected.

Conclusions CD40 rs4810485 SNP was found to confer increased susceptibility to SLE and RA in a Greek population.